Mullerian anomalies and value of diagnosis with 2D ultrasonography

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Abstract

Aim: Mullerian anomalies may accompany some gynecologic, reproductive, or obstetrical problems. They should be kept in mind as a factor for infertility and poor obstetrical results. Conventionally, 2D ultrasonography has been used for diagnosing mullerian anomalies. We evaluated mullerian anomalies diagnosed in our clinic and investigated the prediction of them by 2D ultrasonography. Material and Method: In this study, 82 patients were included who had mullerian duct anomalies. We evaluated all the patients through their medical records and interventions such as 2D ultrasonography, laparoscopy, and hysteroscopy. Results: Out of 82 patients, 53 suffered from infertility. Of the infertile patients, 67.9% (36/53) had uterus septus. The mean duration of infertility of the cases was 2.76 years (±SD 3.27). When we compared ultrasonography against laparoscopy findings, the sensitivity of ultrasonography was 96% and specificity was 15%. Also, comparing ultrasonography against hysteroscopy in diagnosis, the sensitivity and positive predictive value of ultrasonography were both 90%. But specificity was 33%. Discussion: The results of this study suggest that, laparoscopy and hysteroscopy is the gold standard for the diagnosis of mullerian anomalies when compared to 2D ultrasonography, as specificity is low with ultrasonography.

Keywords

Mullerian Anomalies; Ultrasonography; Laparoscopy; Hysteroscopy.

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Introduction

Mullerian anomalies are a common problem in female reproductive tract development [1]. Although the true prevalence is unknown, some studies have reported a prevalence in the range of 0.16% to 10% [1]. Mullerian duct anomalies comprise abnormalities of the uterus, cervix, fallopian tubes, and vagina. These abnormalities can lead to gynecologic, fertility, or obstetrical problems. Some anomalies can be diagnosed in childhood, whereas others can only be detected incidentally, at the time of a clinical evaluation or surgical procedures for other medical conditions [2]. In diagnosis, although hysterosalpingography, ultrasonography, magnetic resonance imaging (MRI), laparoscopy, and hysteroscopy may be used, hysteroscopy and laparoscopy are the gold standards in the diagnosis and treatment of mullerian anomalies [3]. Women who have mullerian duct anomalies present with higher rates of spontaneous abortion, premature delivery, abnormal fetal presentation, and dystocia [1]. Therefore, mullerian anomalies should be kept in mind as a factor for infertility and poor obstetric results, which may require intervention during the pediatric, adolescent, and reproductive years. Simple diagnostic procedures such as pelvic or vaginal 2D ultrasonography are crucial for suspected mullerian duct anomalies at first examination.

Material and Method

Patients who were admitted to our clinic and diagnosed with mullerian anomalies from 2010 to 2015 were included in this study. Most of these patients suffered from infertility and obstetrical problems (e.g., habitual abortus). All cases were evaluated by anamnesis, gynecologic examination, and ultrasonography. 2D Ultrasonography was performed with a Honda Convex Scanner (model HS-2000) and a vaginal probe at 5.0 MHz. Afterwards, laparoscopy and hysteroscopy were performed in all patients under intravenous general anesthesia and using a 5 mm hysteroscope tipped with a 30 degree lens with incorporated 1.5 mm working channel and laparoscopy optic with a 0 degree lens (Karl Storz, Tuttlingen, Germany). Normal saline distension medium was used by a peristaltic irrigating suction device for hysteroscopy. Endouterine pressure was always set below 120 mmHg in hysteroscopy and intraabdominal pressure was set to 14 mmHg in laparoscopy. All hysteroscopic and laparoscopic interventions were recorded by video camera and the whole inspection and operative procedures were noted for each patient's medical record. Hysteroscopy features were classified as normal uterus, septate uterus, uterus bicollis and septate uterus, septate uterus and vagina, transverse septate vagina, or unicornuate uterus. Laparoscopic findings were classified as normal uterus, uterus bicornuate, uterus unicornuate, or uterus didelphys. We also noted the presence of endometriosis. 2D Ultrasonography images were classified as normal uterus, septate uterus, uterus bicornuate, uterus unicornuate, and uterus didelphys. All medical records were kept using the program Microsoft Office Excel 2007 and SPSS 15.0 for Windows Evaluation Version was used in statistical analysis.

Results

Eighty-two patients were enrolled in the study. The age of the patients ranged from 14 to 53 years, with a median of 28.00 years. The age of menarche ranged from 10 to 16 years, with a median of 13.00 years. The mean duration of marriage was 7.06 years (±SD 6.65) and the mean infertility time of the patients was 2.76 years (±SD 3.27). Fifty-three patients suffered from infertility and 67.9% (36/53) of infertile patients had uterus septus.

After gynecologic examination, transverse vaginal septum 8.5% (7/82), longitudinal vaginal septa 11% (9/82), bicollis 15.9% (13/82), and unicollis 84.1%(69/82) were diagnosed. Evaluation of transvaginal sonography revealed septate uterus 35.4% (29/82), uterus didelphys 8.5% (7/82), uterus bicornuate 39.0% (32/82), uterus unicornuate 6.1% (5/82), and normal 11.0 (9/82). Twenty patients (24.4%) suffered from abortus imminens and habitual abortus before their pregnancy. Laparoscopic evaluation revealed bicornuate uterus 15.9% (13/82), uterus didelphys 15.9% (13/82), unicornuate uterus 6.1% (5/82), and normal uterus 62.2% (51/82) (Table 1). Endometriosis was diagnosed in 43.9% (36/82) of patients by laparoscopy. Hysteroscopy revealed 62.2% (51/82) uterus septus, 7.3% (6/82) uterus septus with bicollis, 9.8% (8/82) longitudinal vaginal septum with uterus septus, 4.9% (4/82) transverse vaginal septum, 4.9% (4/82) unicornuate uterus, and 11% (9/82) normal uterus (Table 1). When we compared the findings of ultrasonography and laparoscopy, the sensitivity of ultrasonography was 96% (Table 2). Also, comparing ultrasonography with hysteroscopy in diagnosis, the sensitivity and positive predictive value of ultrasonography were both 90% (Table 2).

Table 1. Diagnosed mullerian abnormalities

	n	%
Bicornuate uterus	13	15.9
Uterus didelphys	13	15.9
Unicornuate uterus	5	6.1
Septate uterus	51	62.2
Septate uterus+Bicollis uterus	6	7.3
Longitudinal vaginal septum with uterus septus	8	9.8
Transverse vaginal septum	4	4.9

Table 2. (A) Comparison of ultrasonography against laparoscopy, (B) comparison of ultrasonography against hysteroscopy.

	A	B
Sensitivity	96%	90%
Specificity	15%	33%
Positive predictive values	41%	90%
Negative predictive values	88%	22%

Discussion

The etiology of mullerian agenesis is currently unknown [3]. One of hypotheses is activation of antimullerian hormone in the genetically female fetus and regression of the mullerian duct [3]. Some studies have reported a prevalence ranging from 0.16% to 10% [1]. We found that the prevalence of mullerian agenesis was 0.25% in our clinic.

The most frequently seen mullerian anomaly is septate uterus [4], as a consequence of incomplete resorption of the uterovaginal septum after fusion of the paramesonephric ducts. These

anomalies are associated with recurrent spontaneous pregnancy losses and adverse obstetrical outcomes, such as preterm birth. Recurrent spontaneous pregnancy losses ranges from 26% to 94% [1]. It has been suggested that the spontaneous abortion rate is reduced from 88% to 5.9% after hysteroscopic resection of the septum [4].

Bicornuate uterus patients have little difficulty conceiving. Spontaneous abortion rates are reported to range from 28% to 35% [5]. The rates of spontaneous abortion and premature delivery depend on the degree of nonfusion of the horns [6]. Bicornuate uterus has been reported to have the highest associated prevalence (38%) of cervical incompetence among mullerian duct anomalies [7]. These patients may require close monitoring during pregnancy and prophylactic replacement of a cervical cerclage in selected patients has been reported to increase fetal survival rates [8].

Uterus didelphys is complete uterine nonfusion. In this pathology, two separate uterine hemicavities either share a cervix or each has its own cervix. There is no communication between the duplicated endometrial cavities [9]. A longitudinal vaginal septum is associated in 75% of these anomalies [10]. Spontaneous abortion rates range from 32% to 52% [4].

Endometriosis and pelvic adhesions have an increased prevalence as a result of retrograde menstrual flow in mullerian anomaly [11]. In our study, 43.9% (36/82) of patients were diagnosed with endometriosis by laparoscopy.

Unicornuate uterus may present as a totally isolated anomaly, but usually presents with a non-communicating rudimentary horn [9]. Unicornuate uterus is associated with spontaneous pregnancy losses and preterm labor [2]. Spontaneous abortion rates are reported to range from 41% to 62% [5]. Usually, in this anomaly, the uterine cavity is only large enough for one fetus and multifetal pregnancy increases the risk for preterm labor [12]. Gestational capacity has been observed to be proportional to uterine muscular organ mass [12]. Resection of the non-communicating horn is indicated in dysmenorrhea with hematometra and also, ectopic pregnancy may occur in the rudimentary horn by means of transperitoneal sperm migration [13]. Renal abnormalities are more commonly associated with unicornuate uterus than with other mullerian duct anomalies and have been reported in 40% of these patients [14].

Transverse vaginal septum can occur anywhere along the vagina, although it occurs most frequently at the junction of the upper and middle third [15]. It causes hematocolpos in patients with a uterus with functioning endometrium [15]. Imperforate hymen is not a mullerian anomaly and should be distinguished from a transverse vaginal septum [2,15]. Longitudinal septa are most often associated with septate and duplication anomalies [15]. When a vaginal obstruction occurs, it is usually unilateral [15].

The woman with mullerian agenesis has normal external genitalia, normal secondary sexual characteristics, and normal karyotypes [16]. Mullerian agenesis is generally diagnosed at puberty because of primary amenorrhea. If endometrial tissue is present in the rudimentary uterine horns, the patient may complain of primary amenorrhea associated with cyclical lower abdominal pain [3].

Mullerian abnormalities are generally diagnosed by radiological (ultrasound, hysterosalpingography, and MRI) or endoscopic examination. Vaginal examination reveals short or absent vagina, transverse or longitudinal vaginal septum, and duplicated cervix [3]. The main differential diagnosis of mullerian agenesis is testicular feminisation syndrome, whose karyotype in this condition is 46, XY [3].

Laparoscopy and hysteroscopy are preferred for the definitive diagnosis, because 2D ultrasonography is not enough to clearly identify the uterus or mullerian rudiments [17]. Also, specificity is low and, at the same time, treatment may accompany diagnosis during laparoscopy and operative hysteroscopy. Especially in our study, we found septate uterus in infertile couples and in the same procedure, treated by hysteroscopy. When we compared the ultrasonography with laparoscopy and also with hysteroscopy, the specificity was 15% and 33%, respectively. On the other hand, effectiveness of 3D ultrasonography on diagnosing of mullerian anomalies has been reported to have over 90% sensitivity and specificity [18-19]. It is an accurate and non-invasive technique to detect and classify uterine mullerian anomalies but its accuracy is related to physician's experience. Additionally, physicians often have easier access to 2D ultrasonography than to 3D ultrasonography.

MR imaging is considered the ideal imaging modality for evaluation of mullerian anomalies. MR imaging provides clear anatomic detail of both the internal uterine cavity and the external contour. Sensitivity of MR imagining has been reported as 77-79% [20-21]. This low sensitivity results from inadequate experience of the radiology physicians for diagnosis and there are no well-established accurate diagnostic criteria based on MR images for mullerian anomalies. Besides, cost is a restrictive factor for extensive usage of MR imaging.

A combined hysteroscopic and laparoscopic evaluation of uterine morphology is considered to be the gold standard method in differential diagnosis of mullerian anomalies. Despite its low sensitivity and specificity, 2D ultrasonogaphy is a more feasible, accessible, and simple procedure. In particular, mullerian anomalies which were suspected after 2D ultrasonography examination in infertile patients should be confirmed by hysteroscopic and laparoscopic evaluation. In this way, the opportunity is provided for correction of some anomalies at the same time as diagnosis.

Scientific Responsibility Statement

The authors declare that they are responsible for the article's scientific content including study design, data collection, analysis and interpretation, writing, some of the main line, or all of the preparation and scientific review of the contents and approval of the final version of the article.

Animal and human rights statement

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. No animal or human studies were carried out by the authors for this article.

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Conflict of interest

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